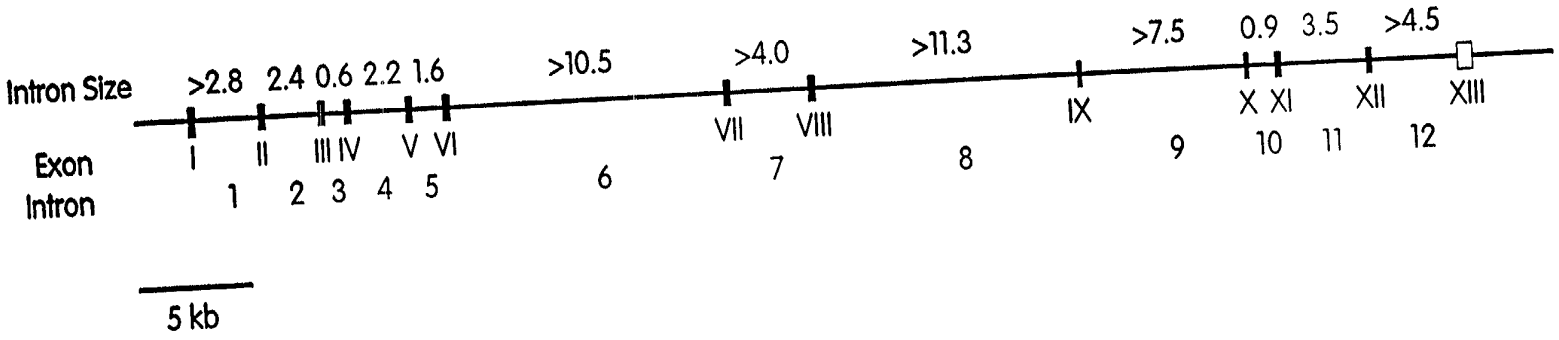

Fig. 1

#5

promoter and exon 1

ACTGCGGAGATGAGGGTCTAGAAGGTGGTGGCGGGGCAT
GTGGACCGTTGTAAGGGCTCTGGGG**TTCCTGGGTGGGCT**
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CCCGATAGAGGAGGAGAGGGAGGAGGAGGGAAAAGGAAG
GGTGAGGGGCTCAGAGGGGAGAGCTGGGAGGAGGGGAGA
CATAGGTGGGGGAAGGGGTAGGAGAAAGGGGAAGGGAGC
AAGAGGGTGAGGGGCACCAGGCCCCATAGACGTTTTGGC
TCAGCGGCCACGAGG**CTTCATCAGCTCCCGCCCGAAAAC**
GGAAGCGAGGCCGTGGGGGCAGCGGCAGCATGGCGGGGC
TTGTCTTGGCGGCCATGGCCCCGCCCCCTGCCCGTCCGA
TCAGCGCCCCGCCCCGTCCCCGCCCCGACCCCGCCCCGG
GCCCGCTCAGGCCCCG**CCCCTGCCGCCGGAATCCTGAAG**
CCCAAGGCTGCCCGGGGGCGGTCCGGCGGCGCCGGCGAT
GGGGC*ATAAAA*CCACTGGCCACCTGCCGGGCTGCTCC

*TGCGTGCGCTGCCGTCCCGGATCC*ACCGTGCCTCTGCGG
CCTGCGTGCCCGGAGTCCCCGCCTGTGTCGTCTCTGTCG
CCGTCCCCGTCTCCTGCCAGGCGCGGAGCCCTGCGAGCC
GCGGGTGGGCCCCAGGCGCGCA**GACATGGGCTGCTCCGC** **A**
CAAAGCGCGCTGGGCTGCCGGGGCGCTGGGCGTCGCGGG
GCTACTGTGCGCTGTGCTGGGCGCTGTCATGATCGTGAT
GGTGCCGTCGCTCATCAAGCAGCAGGTCCTTAAG

GTGGGTGAGGGAGACCCCAGGGGGTCCGCGCACGGACCC
GGGCTGTTGGGCGCTGGGCGCCGGGAGGACCCGCGCGTT
GCGGTGGGTGGGCGACCGCAGCGGAATCGGCGCCCGGGC
CTGGCGCCGCAGAACACGAGGGAGGCCAGGCGCTTCGGG
AGGGGCTGCTGCCCGCCTCCCCACCACCCTCACC

Fig. 2A

exon 2

AGCCTCATGTGCGAAGGGCTTTCCCACCACCTCCTATCC
CAAGCTCCCGCCGAGGAGCCCCTTCCCTGGCCGGGCTCG
GGCAGCTGTTCCGGAGCCTTGTGGTGGGGCG**TGGGGCC**
CTCATCACTCTCCTCACAAGCGTACTTGTCCCTTCCC
CTGCAG

AACGTGCGCATCGACCCCAGTAGCCTGTCCTTCAACATG
TGGAAGGAGATCCCTATCCCCTTCTATCTCTCCGTCTAC
TTCTTTGACGTCATGAACCCCAGCGAGATCCTGAAGGGC
GAGAAGCCGCAGGTGCGGGAGCGCGGGCCCTACGTGTAC
AG

GTGAGGCTGTGTCCACGTGATGGTGGACGGGCCGGCTGA
CGCTGGGCATGGGACGGGTCTCAN**AGTGGACGGGATG**
GGGAGGCTGCTGACTGACCCCCAAACATTGTTCCGGAA
GCACGCAACTCATAGTCGGGGTAAGTGCTACTCCCAAAA
AAGTTTGCGT

exon 3

CATGTCCTGCAGTGGGCAGGCAGCGGGAGGGACAGACTT
GGCGAAGGGGCCGAGCTCAGCTTTGGCTGTGGGGCCGGA
GGTGTGCACAGACGTCCAGGGCCCCTGGTTCCCAGGCAG
GC**ATTGCAGGCGAGTAGAAG**GGAAACGTCCCATGCAG
CGGGGCGGGGCGTCTGACCCACTGGCTTCCCCCACAG

GGAGTTCAGGCACAAAAGCAACATCACCTTCAACAACAA
CGACACCGTGTCCTTCCTcGAGTACCGCACCTTCCAGTT
CCAGCCCTCCAAGTCCCACGGCTCGGAGAGCGACTACAT
CGTCATGCCCAACATCCTGGTCTTG
A
GTGAGGCTGCCCTGTGGCCCACGCCGCCTCGCACCCTGA
CCTCGTCCCC**TGTCTCTCCTCCCGCCTG**CCCCTTGTG
CAGAGAGCAGTCCCTGAGGTGGTCGGAGCGTGGGGACTC
ACGCCTGGTGGGTGGCTTTCGGCCCTGTGCTGTCTCCAC
CACCCCCA

Fig. 2B

exon 4

GGTGGTTCTGGTGTCCCAGATGCCCCACGTGGCCACTCC
AGGGGCCTCCTGCACCCCAGCATTTCCCTTCA**TGGGCT**
CTTTGCTGTGAGGCCCAGCTGGGGCCAAGGGAGGATG
GGCCAGCCACGTCCAGCCTCTGACACTAGTGTCCCTTCG
CCTTGCAG

GGTGCGGCGGTGATGATGGAGAATAAGCCCATGACCCTG
AAGCTCATCATGACCTTGGCATTCACCACCCTCGGCGAA
CGTGCCTTCATGAACCGCACTGTGGGTGAGATCATGTGG
GGCTACAAGGACCCCTTGTGAATCTCATCAACAAGTACT
TTCCAGGCATGTTCCCCTTCAAGGACAAGTTCGGATTAT
TTGCTGAG

GTACGTGTGGCCTGGTGAGAAGCCAAAGATTCAGGCCTG
TGTCCTGT**CTTCCCCTCACACAGCCTGG**ACACTGGTC
ACCAGCTTGCTTTGTAGCTGGCTGGGGATCTAGTGGCTG
TGGGTTGTAAGTGACTGAGAACCTGACTCAAACCGGCTT
GAGTGAAA

exon 5

CCTCTCGGTCCCCAGACACTGGGCATTTGGCAGTGAACC
AGATGCTGGGGGCCCTGTCCTTCTGGTGGAGGGGGAGGA
GGGCTCA**GCCCAGAATGTTCAGACCAG**GCCGGCTCAA
TGGCAGGCCTAAGCCTTACGATGCTGTTCCCTGCTGTGT
CTGTAG

CTCAACAACTCCGACTCTGGGCTCTTCACGGTGTTCACG
GGGGTCCAGAACATCAGCAGGATCCACCTCGTGGACAAG
TGGAACGGGCTGAGCAAG

GTGAGGGGCGAGAGGCGAGGGCCCCTGTCGCCAGGGAGA
GGGGAGGGTGGGCC□GGCCATGGCTGCTCGGGAGTGGCA **T**
GGGACCAGAGAGCTCCTTCTTC CTTTGTCGTGAAGAG
GGTGCTGGGAGGATGAACACTCTTGAAGTTGGAGGAGGG
ATTTTA

Fig. 2C

exon 6

TCTCTGTGTGTCTACATAGCCTGCCCTCTTCCCACCGTG
CCAGTATTGGGAATTGAGTGGCCGTGCGTGCACCAGGGT
GAGTTAGGTGTGCAG**CACCTGAGAGGGCTTATTA**AGG
GGCCTTGGCCCTACTGAGGGGTCTAGTCTGGATGCTTCC
CCCCAG

GTTGACTTCTGGCATTCCGATCAGTGCAACATGATCAAT
GGAACTTCTGGGCAAATGTGGCCGCCCTTCATGACTCCT
GAGTCCTCGCTGGAGTTCTACAGCCCGGAGGCCTGCCG

GTAATCACTGGGACTCGGGGCCTCCTGGGTTTCCTGGGT
AGCTCATGGCCAAATTCTGTGGTGTTGGCTGT**GCACTT**
GGAAAGCATTTTGACTCATCGTGGATTTGACTCAGTAG
CCCTTGGCACCAGCTTGAATTCTCTTTGGTCACACCACC
AAAAGC

exon 7

GGAGGTCGCTGCAGCTCCGCGGGTGAGAGATGGGGGCGG
TTTGGACCCGGGAGGTGGTAGCGCCCGTGGGGAGAAGTG
GCTGGATCTGGGCAGCCTTTGGCAGGGCCTGGCTCTG**GC**
CGCCGGGTCTGGGTGTCCCCTCTCATCCTGTCTGTCC
CCTGCAG

ATCCATGAAGCTAATGTACAAGGAGTCAGGGGTGTTTGA
AGGCATCCCCACCTATCGCTTCGTGGCTCCCAAAACCCT
GTTTGCCAACGGGTCCATCTACCCACCCAACGAAGGCTT
CTGCCCGTGCCTGGAGTCTGGAATTCAGAACGTCAGCAC
CTGCAGGTTCA

GTACGTGCCGTCCCCTGTTCTGGGATNGCCGGAGGGTGT
TAGGTNTNGGGCACCTNANGGTTTATCTGCCCAATGCTG
TCTGCTTAATCTCTGGCCTCTGTACTCTTGATAACC
CATTAAGCCAAAAATATGATGCCTCTGGGACGATATCTG

Fig. 2D

exon 8

TGGGGC**TTTTTACAGAATGGAGG**AAGGGATCCTCTCT
GTCGGGTATTATGGTCATCGCCACGGGGGTGCCGTGCAG
ACCACAGCTCTGTGCAGACTTCCGGAGTGGCAGGACGTG
CCAATATACTGTCGTTGTATGATGTCCCCTCCCTGCCCT
TGTTGTAG

GTGCCCCCTTGTTTCTCTCCCATCCTCACTTCCTCAACG
CCGACCCGGTTCTGGCAGAAGCGGTGACTGGCCTGCACC **T**
CTAACCAGGAGGCACACTCCTTGTTCCTGGACATCCACC
CG

GTGAGCCCCTGCCATCCTCTGTGGGGGGTGGGTGATTCC
TGGTTGGAGCACACCTGGCTGCCTCCTCTCTCCCCAG
GCAGAGAGCTGCTGTGGGCTGGGGTGGTGGGAAGCCTGG
CTTCTAGAATCTCGAGCCACCAAAGTTCCTTACT

exon 9

CCCCAGCCTGTGGCTTGTTTTAGGTAAGATACAAGCAAG
CTCCACTGGGCAGTTAGCTGGGACGCCCACCCTCTTGAC
TGGGACCAGGGAAAAGAA**GGTTGACTGTGTCCCTGGA**
GCTTGGGGGTGGCCAGTCTCCTCACTGTGTTTGTTGCCG
CAG

GTCACGGGAATCCCCATGAACTGCTCTGTGAAACTGCAG
CTGAGCCTCTACATGAAATCTGTCGCAGGCATTGG

GTGAGTGGGGACTGGGAACTGGGGCTGCATTGCTCATTG
AGAGATTANGT**GCTCAGTGCTCCAGTGTTCCC**AGAC
TCCCCTGACATACCCCAGGAAACAGGGCATGGGGAAGGG
AGAGGGTCCTATTGGGGGTGGAATCCAGTCCCTGCTGAT
CTTCTC

Fig. 2E

exon 10

ATGGCTCCTAAAGTGTTTCAGCTCATTGTTTATATTT**GG**
TGGTGAGGGTTTAGTGTGTGCAAAATTATACTAAACC
TGTTTAGATGTTGTATTCAAGCAGAATTAGATCAAGTTT
GGGTGTAAGACTTTGTTCCAACACCTATGTCTTGCTTAT
TTCCAG

ACAAACTGGGAAGATTGAGCCTGTGGTCCTGCCGCTGCT
CTGGTTTGCAGAG

GTAAGGGTGCGTTGGGCACAGCGTCGGGGGCTTTTGTTA
ATAGCCAATGTGGGCATTT**GAGGCAGGAGGCGGGGGG**
AGCACCTTGTAGAAAGGGAGAGGGCTGAGCCAGGGTAAC
CGGACTGTTACATGGACCAGCGTATCATACACTTCACCC
TGTC

exon 11

CCTGGAGGGAGGAGGTCCCTGGCAGGCTCCAACACATGC
TTTAGCCGGGAAGCTTGAGGTGGGGAAAAGCTGAGGCGG
GCACAGAGG**AAGGTGTTGGGTGGCATCTG**CGCTGTAG
CCCGCAGCCTGCGGCCCCAGCTCATGTGTTTGTCATTCT **G**
GTCTCCTCAG

AGCGGGGCCATGGAGGGGGAGACTCTTCACACATTCTAC
ACTCAGCTGGTGTTGATGCCCAAGGTGATGCACTATGCC
CAGTACGTCCTCCTGGCGCTGGGCTGCGTCCTGCTGCTG
GTCCCTGTCATCTGCCAAATCCGGAGCCAA

GTAGGTGCTGGCCAGAGGGCAGCCCGGGCTGACAGCCAT
TCGCTTGCCTGCTGGGGGGAAAGGGGCCTCAGATCGGACC
CTCTG**GCCAACCGCAGCCTGGAGCC**CACCTCCAGCAG
CAGTCCTGCGTCTCTGCCGGAGTGGGAGCGGTCACTGCT
GGGGG

Fig. 2F

exon 12

CCCCACATCTCAGCCACCTGCAATCGTTGAGGGTTGTTG
GACTCTAAACTTATGTGCCTTTCCTGTTTCCTCTTTGCC
TTTTGCAAA**TTGAAGAACCGTGTAAAAC**CATTTTTAT
GTGGCTTCAACGTCAACTATAAATTAGCTTGGTTATCTT
CTAG

GAGAAATGCTATTTATTTTGGAGTAGTAGTAAAAAGGGC
TCAAAGGATAAGGAGGCCATTCAGGCCTATTCTGAATCC
CTGATGACATCAGCTCCCAAGGGCTCTGTGCTGCAGGAA
GCAAAACTGTAG

GTGGGTACCAGGTAATGCCGTGCGCCTCCCCGCCCCCTC
CCATATCAAGTAGAATGCTGGCGGCTTAAAACATTTGGG
GTCCTGC**TCATTCCTTCAGCCTCAA**CTTCACCTGGAG
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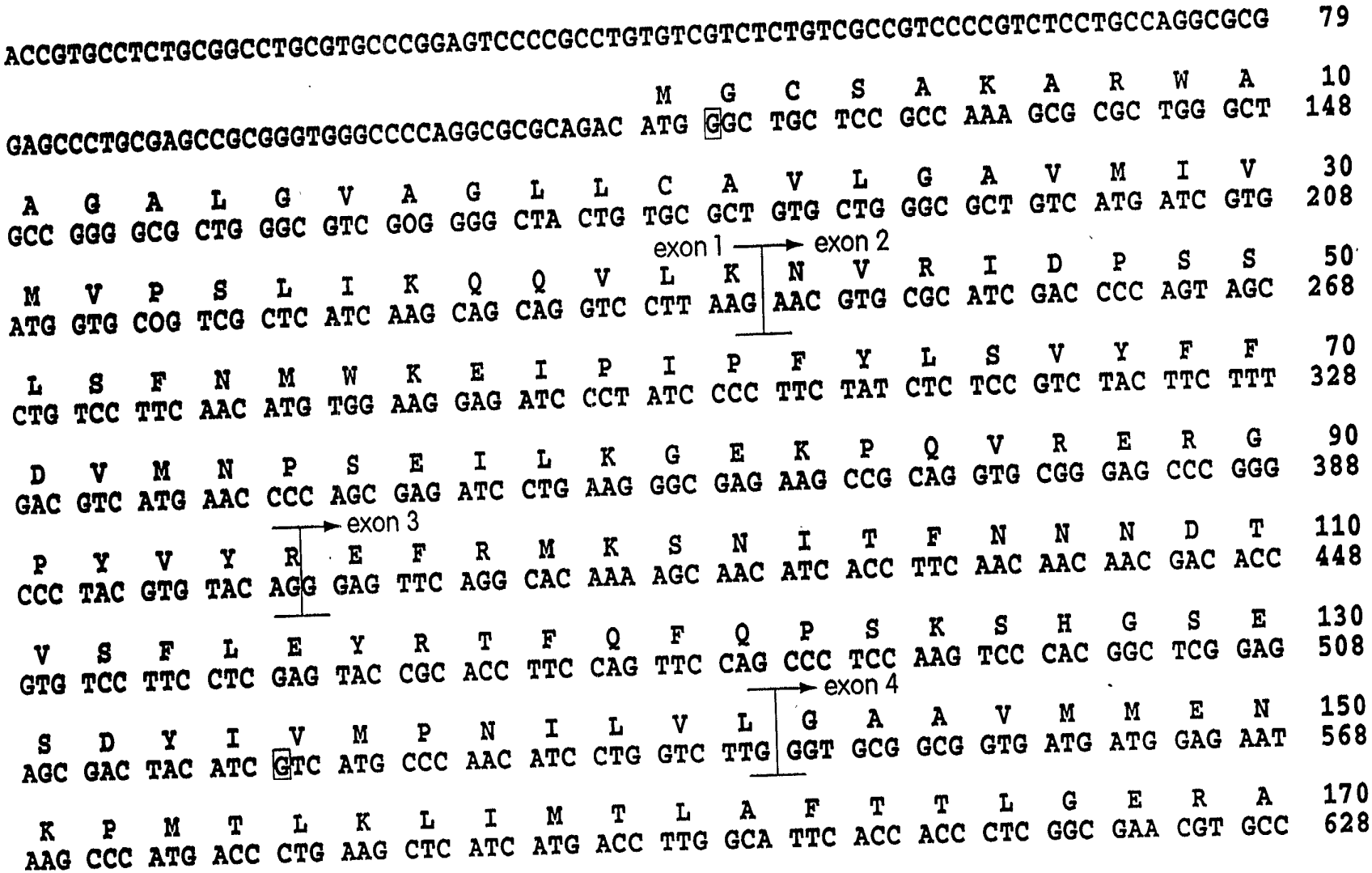
TTGGAGAAA

Fig. 2G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A G A L G V A G L L C A V L G A V M I V 30
GCC GGG GCG CTG GGC GTC GOG GGG CTA CTG TGC GCT GTG CTG GGC GCT GTC ATG ATC GTG 208
exon 1
exon 2
M V P S L I K Q Q V L K N V R I D P S S 50
ATG GTG COG TCG CTC ATC AAG CAG CAG GTC CTT AAG AAC GTG CGC ATC GAC CCC AGT AGC 268
L S F N M W K E I P I P F Y L S V Y F F 70
CTG TCC TTC AAC ATG TGG AAG GAG ATC CCT ATC CCC TTC TAT CTC TCC GTC TAC TTC TTT 328
D V M N P S E I L K G E K P Q V R E R G 90
GAC GTC ATG AAC CCC AGC GAG ATC CTG AAG GGC GAG AAG CCG CAG GTG CGG GAG CCC GGG 388
exon 3
P Y V Y R E F R M K S N I T F N N N D T 110
CCC TAC GTG TAC AGG GAG TTC AGG CAC AAA AGC AAC ATC ACC TTC AAC AAC AAC GAC ACC 448
V S F L E Y R T F Q F Q P S K S H G S E 130
GTG TCC TTC CTC GAG TAC CGC ACC TTC CAG TTC CAG CCC TCC AAG TCC CAC GGC TCG GAG 508
exon 4
S D Y I V M P N I L V L G A A V M M E N 150
AGC GAC TAC ATC GTC ATG CCC AAC ATC CTG GTC TTG GGT GCG GCG GTG ATG ATG GAG AAT 568
K P M T L K L I M T L A F T T L G E R A 170
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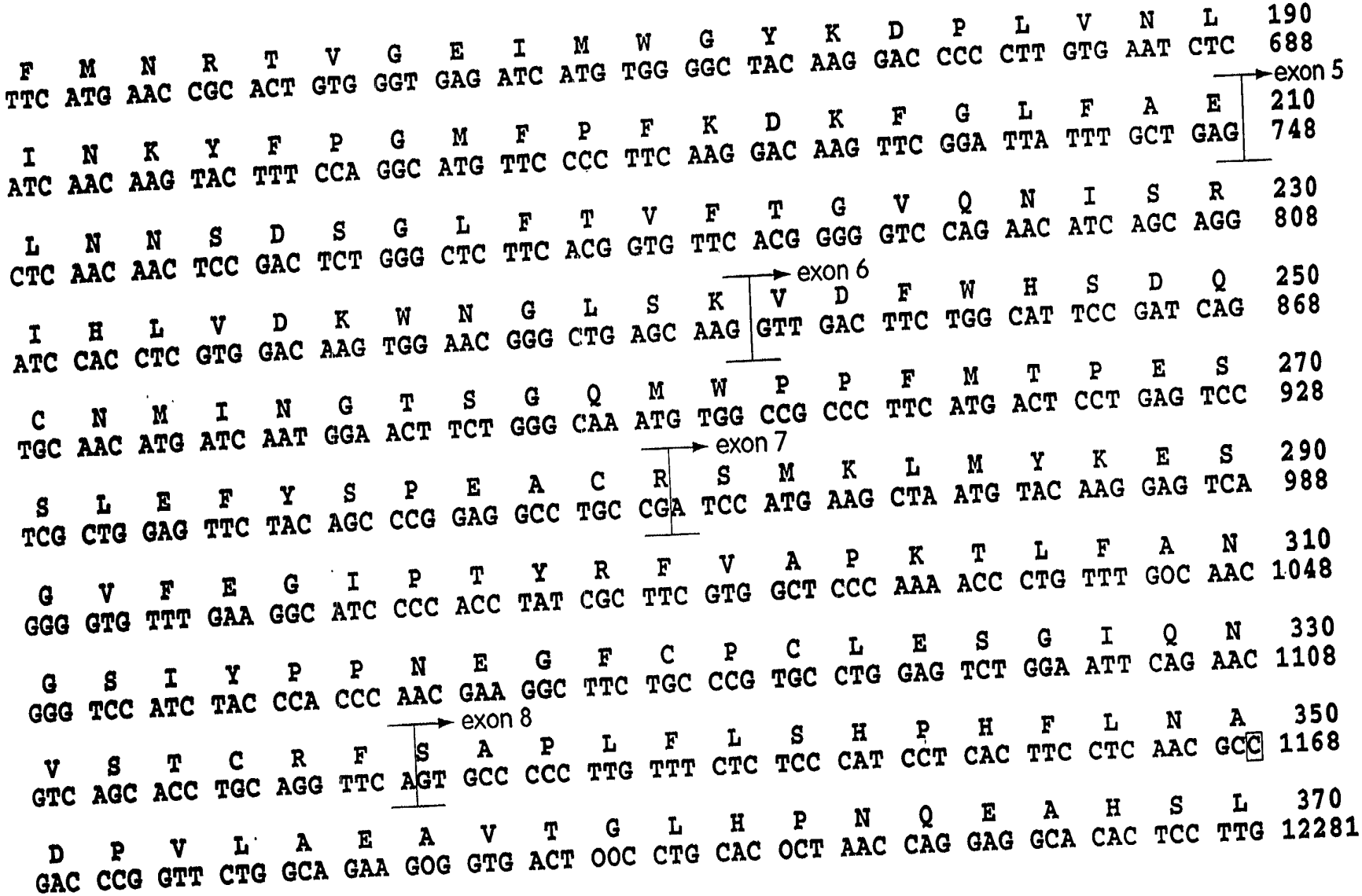
Fig. 3A-1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exon 5
I N K Y F P G M F P F K D K F G L F A E 210
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L N N S D S G L F T V F T G V Q N I S R 230
CTC AAC AAC TCC GAC TCT GGG CTC TTC ACG GTG TTC ACG GGG GTC CAG AAC ATC AGC AGG 808
exon 6
I H L V D K W N G L S K V D F W H S D Q 250
ATC CAC CTC GTG GAC AAG TGG AAC GGG CTG AGC AAG GTT GAC TTC TGG CAT TCC GAT CAG 868
C N M I N G T S G Q M W P P F M T P E S 270
TGC AAC ATG ATC AAT GGA ACT TCT GGG CAA ATG TGG CCG CCC TTC ATG ACT CCT GAG TCC 928
exon 7
S L E F Y S P E A C R S M K L M Y K E S 290
TCG CTG GAG TTC TAC AGC CCG GAG GCC TGC CGA TCC ATG AAG CTA ATG TAC AAG GAG TCA 988
G V F E G I P T Y R F V A P K T L F A N 310
GGG GTG TTT GAA GGC ATC CCC ACC TAT CGC TTC GTG GCT CCC AAA ACC CTG TTT GOC AAC 1048
G S I Y P P N E G F C P C L E S G I Q N 330
GGG TCC ATC TAC CCA CCC AAC GAA GGC TTC TGC CCG TGC CTG GAG TCT GGA ATT CAG AAC 1108
exon 8
V S T C R F S A P L F L S H P H F L N A 350
GTC AGC ACC TGC AGG TTC AGT GCC CCC TTG TTT CTC TCC CAT CCT CAC TTC CTC AAC GCC 1168
D P V L A E A V T G L H P N Q E A H S L 370
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Fig. 3A-2

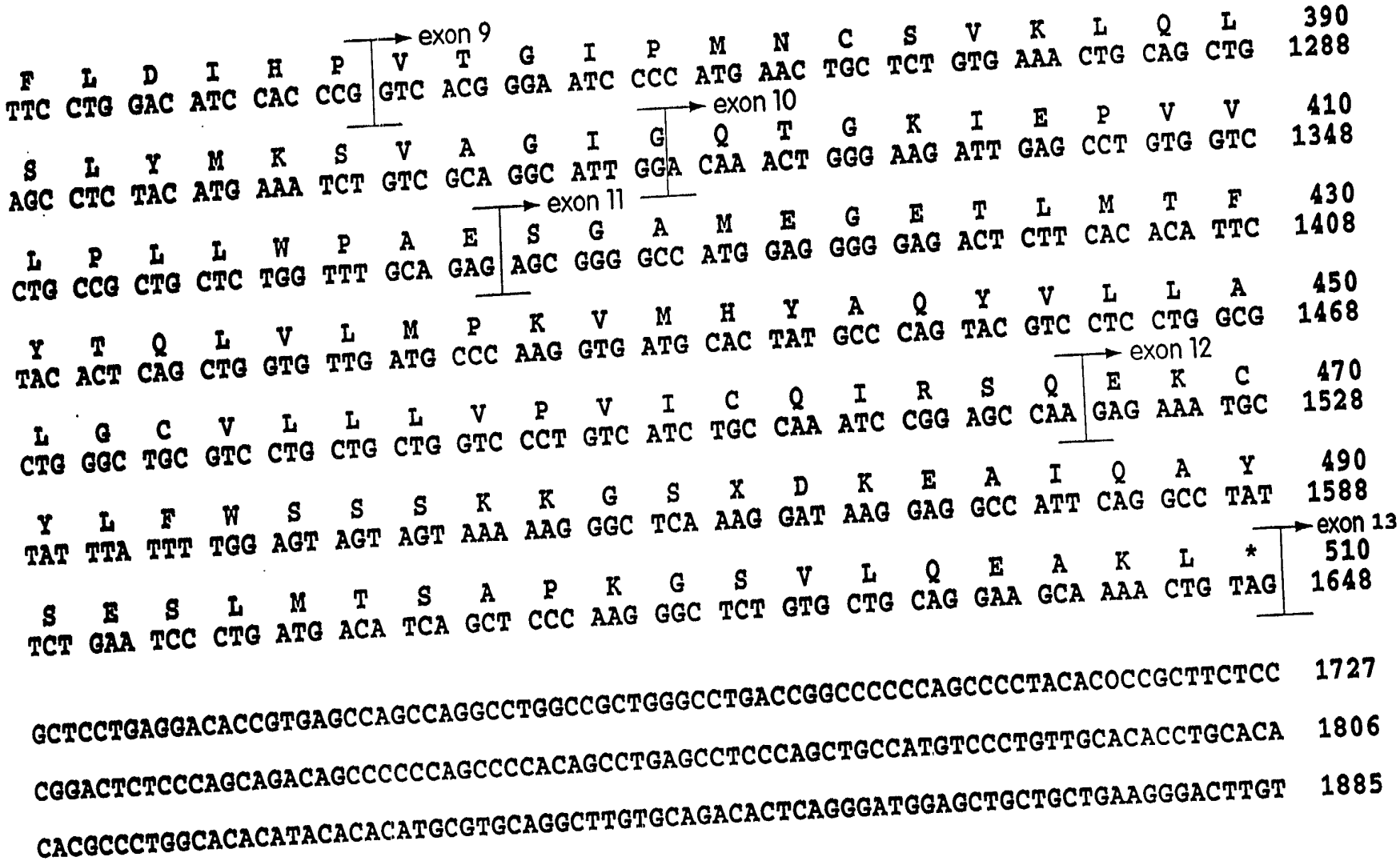

Fig. 3B-1

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GCCTCGGAAAACAATGGAGTGAGCACAAGATGCCCTGTGCAGCTGCCCGAGGGTCTCCGCCCACCCCGGCCGGACTTTG 2280

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GGACTGAGCCGGCCACTYTYTGGCCGAAGTGGCCGCAGGCTGTGCCCCCGAGCTGCCCCCACCCCCTCACAGGGTCCCT 2438

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12/18

Fig. 3B-2

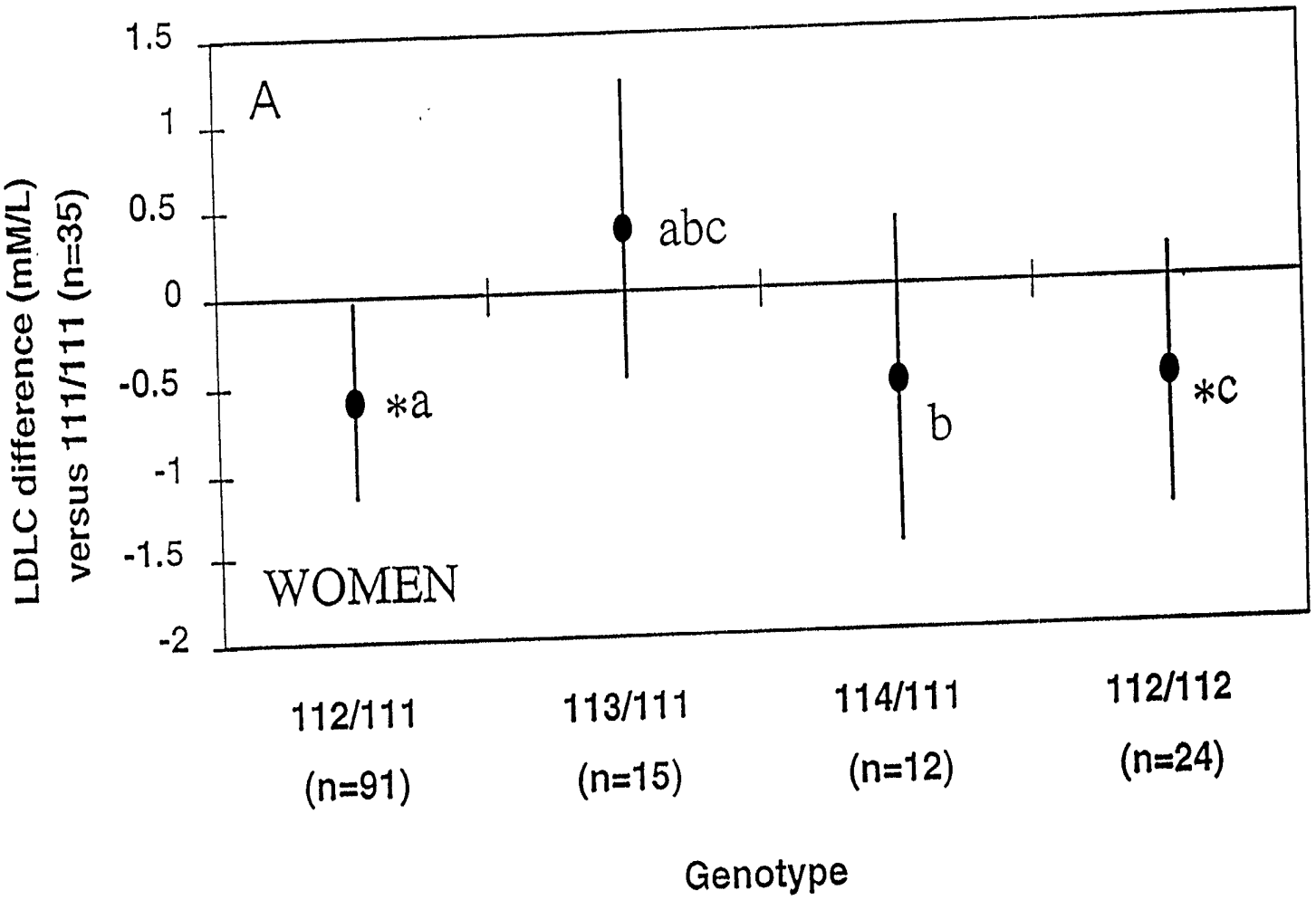
1.5
1
0.5
0
-0.5
-1
-1.5
-2
A
WOMEN
*a
abc
b
*c
LDLC difference (mM/L)
versus 111/111 (n=35)
112/111
(n=91)
113/111
(n=15)
114/111
(n=12)
112/112
(n=24)
Genotype

Fig. 4

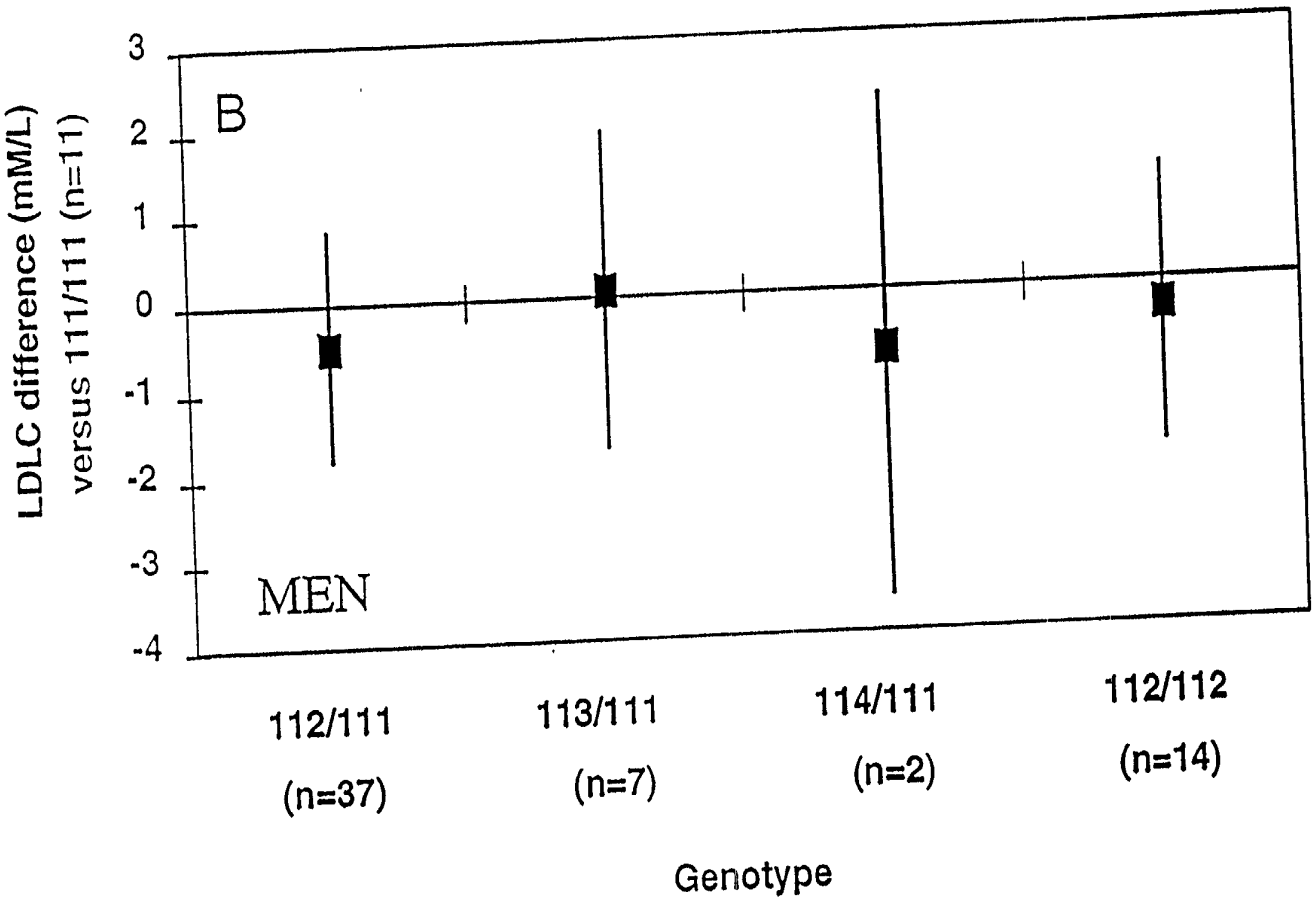
LDLC difference (mM/L)
versus 111/111 (n=11)
3
2
1
0
-1
-2
-3
-4
B
MEN
112/111
(n=37)
113/111
(n=7)
114/111
(n=2)
112/112
(n=14)
Genotype

Fig. 5

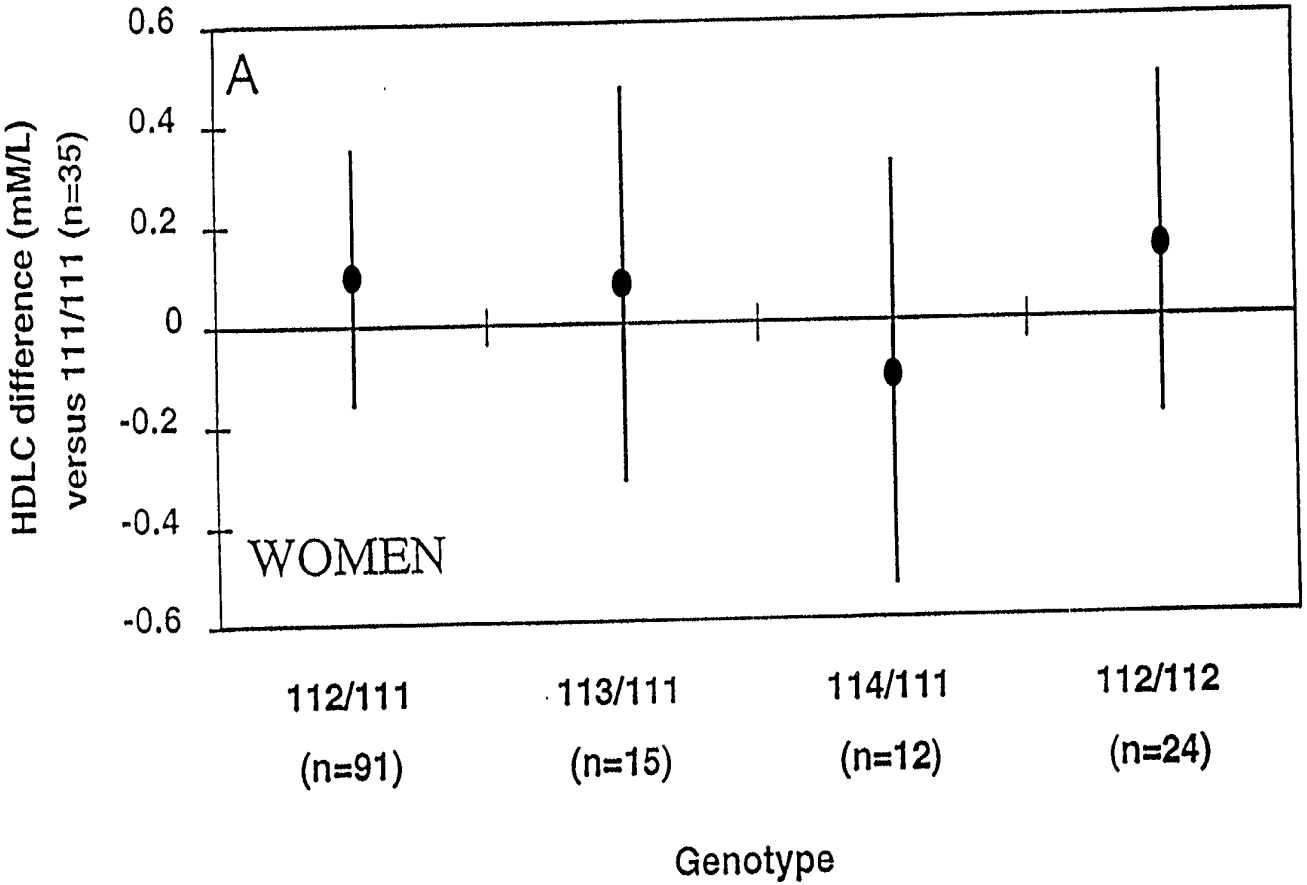
HDLC difference (mM/L)
versus 111/111 (n=35)
0.6
0.4
0.2
0
-0.2
-0.4
-0.6
A
WOMEN
112/111
(n=91)
113/111
(n=15)
114/111
(n=12)
112/112
(n=24)
Genotype

Fig. 6

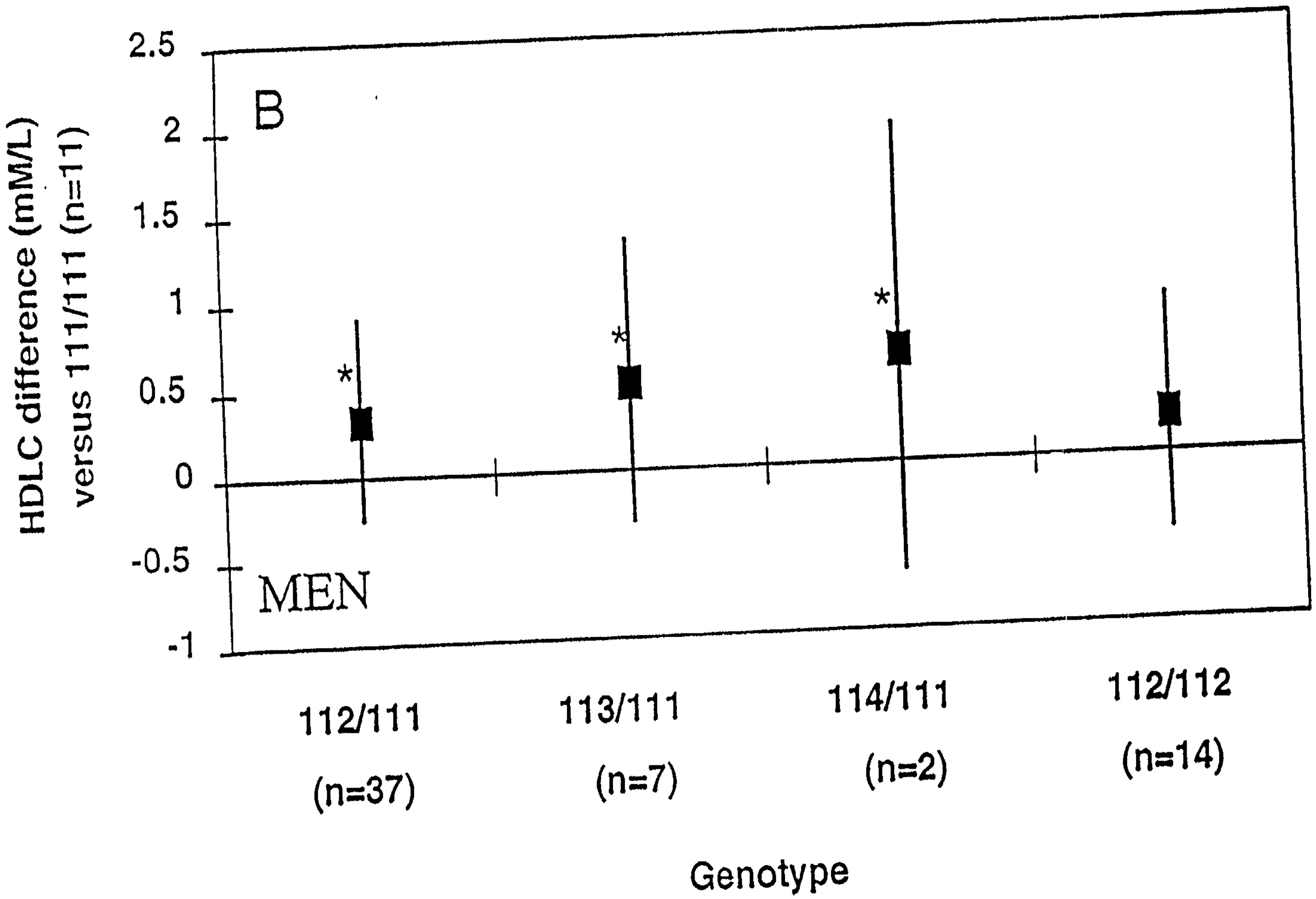

Fig. 7

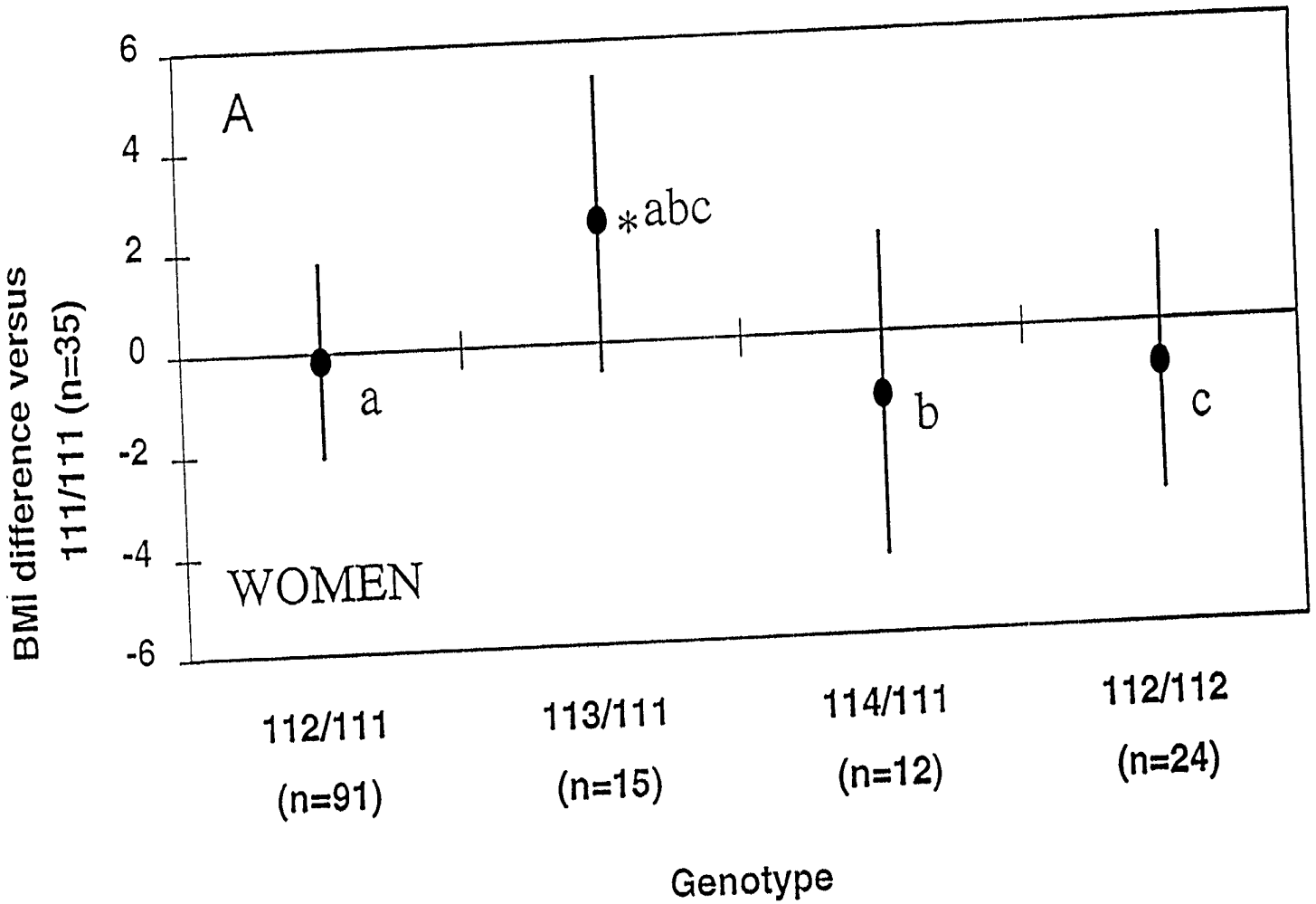
A
WOMEN
BMI difference versus 111/111 (n=35)
6
4
2
0
-2
-4
-6
*abc
a
b
c
112/111 (n=91)
113/111 (n=15)
114/111 (n=12)
112/112 (n=24)
Genotype

Fig. 8

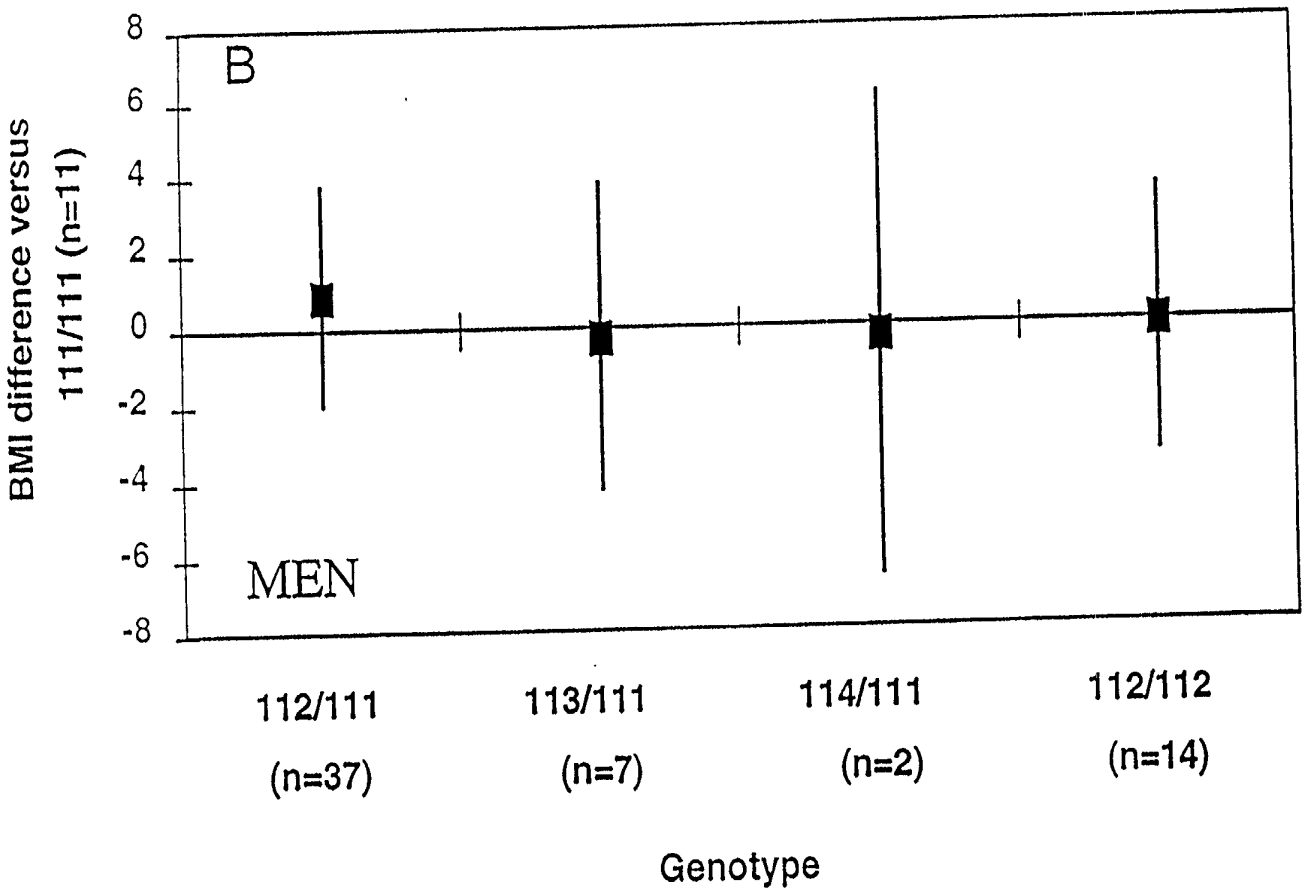
B
MEN
BMI difference versus 111/111 (n=11)
8
6
4
2
0
-2
-4
-6
-8
112/111 (n=37)
113/111 (n=7)
114/111 (n=2)
112/112 (n=14)
Genotype

Fig. 9